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☐ 1: NM_000485. Reports Homo sapiens aden...[gi:71773149]

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<u>Comment</u>	<u>Features</u>	<u>Sequence</u>
LOCUS	NM_000485	807 bp mRNA linear PRI 22-JUN-2008
DEFINITION	Homo sapiens adenine phosphoribosyltransferase (APRT), transcript variant 1, mRNA.	
ACCESSION	NM_000485	
VERSION	NM_000485.2 GI:71773149	
KEYWORDS	.	
SOURCE	Homo sapiens (human)	
ORGANISM	Homo sapiens	
	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo.	
REFERENCE	1 (bases 1 to 807)	
AUTHORS	Silva,C.H., Silva,M., Iulek,J. and Thiemann,O.H.	
TITLE	Structural complexes of human adenine phosphoribosyltransferase reveal novel features of the APRT catalytic mechanism	
JOURNAL	J. Biomol. Struct. Dyn. 25 (6), 589-597 (2008)	
PUBMED	18399692	
REMARK	GeneRIF: Data indicates that the flexible loop structure adopts an open conformation before and after binding of both substrates adenine and phosphoribosyl pyrophosphate.	
REFERENCE	2 (bases 1 to 807)	
AUTHORS	Di Pietro,V., Perruzza,I., Amorini,A.M., Balducci,A., Ceccarelli,L., Lazzarino,G., Barsotti,P., Giardina,B. and Tavazzi,B.	
TITLE	Clinical, biochemical and molecular diagnosis of a compound homozygote for the 254 bp deletion-8 bp insertion of the APRT gene suffering from severe renal failure	
JOURNAL	Clin. Biochem. 40 (1-2), 73-80 (2007)	
PUBMED	17126311	
REMARK	GeneRIF: APRT assay in a sample of patient hemolysate showed no detectable activity of the enzyme (25.56+/-9.55 U/L red blood cells in control healthy subjects).	
REFERENCE	3 (bases 1 to 807)	
AUTHORS	Ewing,R.M., Chu,P., Elisma,F., Li,H., Taylor,P., Climie,S., McBroom-Cerajewski,L., Robinson,M.D., O'Connor,L., Li,M., Taylor,R., Dharsee,M., Ho,Y., Heilbut,A., Moore,L., Zhang,S., Ornatsky,O., Bukhman,Y.V., Ethier,M., Sheng,Y., Vasilescu,J., Abu-Farha,M., Lambert,J.P., Duewel,H.S., Stewart,I.I., Kuehl,B., Hogue,K., Colwill,K., Gladwish,K., Muskat,B., Kinach,R., Adams,S.L., Moran,M.F., Morin,G.B., Topaloglou,T. and Figeys,D.	
TITLE	Large-scale mapping of human protein-protein interactions by mass spectrometry	
JOURNAL	Mol. Syst. Biol. 3, 89 (2007)	
PUBMED	17353931	

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REFERENCE 4 (bases 1 to 807)
 AUTHORS Bruneel,A., Labas,V., Mailloux,A., Sharma,S., Royer,N., Vinh,J.,
 Pernet,P., Vaubourdolle,M. and Baudin,B.
 TITLE Proteomics of human umbilical vein endothelial cells applied to
 etoposide-induced apoptosis
 JOURNAL Proteomics 5 (15), 3876-3884 (2005)
 PUBMED [16130169](#)

REFERENCE 5 (bases 1 to 807)
 AUTHORS Taniguchi,A., Tsuchida,S., Kuno,S., Mita,M., Machida,T.,
 Ioritani,N., Terai,C., Yamanaka,H. and Kamatani,N.
 TITLE Identification of two novel mutations in adenine
 phosphoribosyltransferase gene in patients with
 2,8-dihydroxyadenine urolithiasis
 JOURNAL Nucleosides Nucleotides Nucleic Acids 23 (8-9), 1141-1145 (2004)
 PUBMED [15571218](#)
 REMARK GeneRIF: two novel mutations, G133D and V84M, were found in the
 APRT gene in Japanese patients with APRT deficiency

REFERENCE 6 (bases 1 to 807)
 AUTHORS Kamatani,N., Hakoda,M., Otsuka,S., Yoshikawa,H. and Kashiwazaki,S.
 TITLE Only three mutations account for almost all defective alleles
 causing adenine phosphoribosyltransferase deficiency in Japanese
 patients
 JOURNAL J. Clin. Invest. 90 (1), 130-135 (1992)
 PUBMED [1353080](#)

REFERENCE 7 (bases 1 to 807)
 AUTHORS Chen,J., Sahota,A., Laxdal,T., Scrine,M., Bowman,S., Cui,C.,
 Stambrook,P.J. and Tischfield,J.A.
 TITLE Identification of a single missense mutation in the adenine
 phosphoribosyltransferase (APRT) gene from five Icelandic patients
 and a British patient
 JOURNAL Am. J. Hum. Genet. 49 (6), 1306-1311 (1991)
 PUBMED [1746557](#)

REFERENCE 8 (bases 1 to 807)
 AUTHORS Ludwig,H., Kuzmits,R., Pietschmann,H. and Muller,M.M.
 TITLE Enzymes of the purine interconversion system in chronic lymphatic
 leukemia: decreased purine nucleoside phosphorylase and adenosine
 deaminase activity
 JOURNAL Blut 39 (5), 309-315 (1979)
 PUBMED [116697](#)

REFERENCE 9 (bases 1 to 807)
 AUTHORS Holden,J.A., Meredith,G.S. and Kelley,W.N.
 TITLE Human adenine phosphoribosyltransferase. Affinity purification,
 subunit structure, amino acid composition, and peptide mapping
 JOURNAL J. Biol. Chem. 254 (15), 6951-6955 (1979)
 PUBMED [457664](#)

REFERENCE 10 (bases 1 to 807)
 AUTHORS Johnson,L.A., Gordon,R.B. and Emmerson,B.T.
 TITLE Adenine phosphoribosyltransferase: a simple spectrophotometric
 assay and the incidence of mutation in the normal population
 JOURNAL Biochem. Genet. 15 (3-4), 265-272 (1977)
 PUBMED [869896](#)

COMMENT REVIEWED REFSEQ: This record has been curated by NCBI staff. The
 reference sequence was derived from [BM423481.1](#) and [BU507629.1](#).
 On Aug 3, 2005 this sequence version replaced [gi:4502170](#).

Summary: Adenine phosphoribosyltransferase belongs to the
 purine/pyrimidine phosphoribosyltransferase family. A conserved
 feature of this gene is the distribution of CpG dinucleotides. This
 enzyme catalyzes the formation of AMP and inorganic pyrophosphate
 from adenine and 5-phosphoribosyl-1-pyrophosphate (PRPP). It also

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produces adenine as a by-product of the polyamine biosynthesis pathway. A homozygous deficiency in this enzyme causes 2,8-dihydroxyadenine urolithiasis. Two transcript variants encoding different isoforms have been found for this gene.

Transcript Variant: This variant (1) represents the longer transcript and encodes the longer isoform (a).

Publication Note: This RefSeq record includes a subset of the publications that are available for this gene. Please see the Entrez Gene record to access additional publications.
COMPLETENESS: complete on the 3' end.

PRIMARY	REFSEQ_SPAN	PRIMARY_IDENTIFIER	PRIMARY_SPAN	COMP
	1-713	BM423481.1	25-737	
	714-807	BU507629.1	115-208	

FEATURES	Location/Qualifiers
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<u>exon</u>	1..115 /gene="APRT" /inference="alignment:Splign" /number=1
<u>STS</u>	12..684 /gene="APRT" /db_xref="UniSTS:486660"
<u>CDS</u>	36..578 /gene="APRT" /EC_number="2.4.2.7" /note="isoform a is encoded by transcript variant 1; transphosphoribosidase; AMP pyrophosphorylase; AMP diphosphorylase; adenine phosphoribosyltransferase, isoform a" /codon_start=1 /product="adenine phosphoribosyltransferase isoform a" /protein_id="NP_000476.1" /db_xref="GI:4502171" /db_xref="CCDS:CCDS32511.1" /db_xref="GeneID:353" /db_xref="HGNC:626" /db_xref="HPRD:00029" /db_xref="MIM:102600" /translation="MADSELQLVEQIRISFPDFPTPGVVFRDISPVLKDPASFRAAIG LLARHLKATHGGRIDYIAGLDSRGFLFGPSLAQELGLGCVLIRKRGKLPGPPTLWASYS LEYGKAELEIQKDALEPGQRVVVVDDLLATGGTMNAACELLGRLQAEVLECVSLVELT SLKGREKLAPVPFFSLLQYE"
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exon /inference="alignment:Splice" /number=2 223..356 /gene="APRT" /inference="alignment:Splice" /number=3
exon 357..435 /gene="APRT" /inference="alignment:Splice" /number=4
exon 436..807 /gene="APRT" /inference="alignment:Splice" /number=5a
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STS 481..643 /gene="APRT" /standard_name="RH68125" /db_xref="UniSTS:27274"
polyA_site 807 /gene="APRT"

ORIGIN

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